DOCUMENT RESUME

ED 289 268 EC 201 208

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TITLE Multidisciplinary Developmental Evaluation Improves Early Diagnosis of Infantile Autism.
PUB DATE Apr 87
NOTE 23p.; Paper presented at the Biennial Meeting of the Society for Research in Child Development (Baltimore, MD., April 23-26, 1987).
PUB TYPE Reports - General (140) -- Speeches/Conference Papers (150)

EDRS PRICE MF01/PC01 Plus Postage.
DESCRIPTORS *Autism; *Clinical Diagnosis; Evaluation; Handicap Identification; *Individual Development; *Interdisciplinary Approach; Language Acquisition; *Pediatrics; *Psychopathology; Social Development; Young Children

ABSTRACT

A life-course perspective of the diagnostic histories of 75 autistic individuals (ages 4-25) was obtained through the use of parent surveys and a review of their charts. The study was made to understand better how children who presented with uneven or unusual behavioral development are identified as developmentally multihandicapped. Areas examined included when parents became concerned about developmental delay, what concerns they expressed, to whom they expressed them, when evaluations were made, what kinds of evaluations were carried out, and which diagnostic models were most effective. The results showed that parents most often first expressed concern to pediatricians, noting both language and social delays. On average, parents first reported concerns around 1-1/2 years of age, began diagnostic evaluations around 2-1/2 years of age, and received diagnoses of autism around 4-1/2 years of age. These findings are discussed in terms of the potential value of early identification and placement into infant and preschool intervention programs. The role of the child's primary care physician in early identification and coordination of diagnostic evaluation is discussed. (Author/VW)

Multidisciplinary Developmental Evaluation Improves Early Diagnosis of Infantile Autism

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Paper presented at the Biennial Meeting
of the
Society for Research in Child Development
Baltimore, MD, April 23-26, 1987

Identifying Developmentally Multihandicapped Children

ABSTRACT

Chart review and parent surveys were obtained for 75 children with autism to gain a life-course perspective of their diagnostic histories. The study was made to understand better how children who presented with uneven or unusual behavioral development are identified as developmentally multihandicapped. We examined when parents became concerned about developmental delay, what concerns they expressed, to whom they expressed them, when evaluations were made, what kinds of evaluations were carried out, and which diagnostic models were most effective. The results showed that parents most often first expressed concern to pediatricians, noting both language and social delays. On average, parents first reported concerns around 1-1/2 years of age, began diagnostic evaluations around 2-1/2 years of age, and received diagnoses of autism around 4-1/2 years of age. These findings are discussed in terms of the potential value of early identification and placement into infant and preschool intervention programs. The role of the child's primary care physician in early identification and coordination of diagnostic evaluation is discussed.

Keywords: Early identification, developmental psychopathology, developmental screening, autism, learning disabilities.

Introduction

Children with multiple developmental disabilites present diagnostic problems for both generalist and specialist clinicians. Of particular concern are children who show delays or failures in more than one domain of behavioral development such as language, social adaptiveness, attention span and activity level, while acheiving normal milestones in physical and motor development. Identifying the problems of such a multihandicapped child is like the parable of blind men feeling an elephant: each touches a different part of the elephant and "sees" a different creature. With a multihandicapped child, the pediatrician may see a patient who is plagued by recurrent ear infections and, perhaps for that reason, language delayed; the neurologist may see someone with essentially normal neurological signs, except for unusual stereotyped motor movements; the speech pathologist may see that the child has poor language pragmatics; the psychologist may see mental retardation; and the child psychiatrist may speculate upon a history of psychic trauma to explain the abberant behavior. If all of these "parts" co-exist in one child, it can be difficult for any one evaluator to "see" a complete picture of the disabilities.

Diagnostic classification of multihandicapping developmental or behavioral conditions such as the pervasive developmental disorders (PDD), attention deficit disorders (ADD), and other specific learning disabilities of cognitive and perceptual-motor development is imprecise. Making diagnoses in these disorders often requires crossing disciplinary boundaries either to rule out a variety of possible causes of any given symptom or to understand fully the panoply of the child's impairments. In the present study, we examined how children with autism (a PDD) were identified and diagnosed. We hope this may suggest how such children can be reliably identified earlier, and thereby treated appropriately.

Early infantile autism (EIA) is a pervasive developmental disorder affecting 5-6 children per 10,000, occurring 4-5 times more frequently in males than females (DSM-III,

1980). Diagnostic criteria include specific qualitative impairments in reciprocal social interaction, verbal and non-verbal communication, and a restricted repertoire of activities and interests. Early symptoms of the disorder--failures in social and language development--often go unrecognized because in isolation they are neither necessarily pathological nor pathognomonic. Symptoms of autism can be mistaken for other disorders such as deafness, hyperactivity, emotional disturbance, language disorder, or mental retardation. In working with such patients for a number of years, we became impressed with differences among those who had and had not received early professional interventions and so have begun to explore ways of improving early identification of the disorder.

The purpose of the present studies was to answer the following questions: 1) At what age did parents first become concerned about delays in their children's development? 2) In what domains of development (social, language, physical) did parents first become concerned? 3) At what age was some developmental disability first confirmed (an initial diagnosis)? 4) Which professionals provided initial diagnostic information to parents? 5) What initial diagnoses were given? 6) How accurate or complete were these diagnoses? 7) When initial diagnoses other than autism were given, at what age was the definitive diagnosis of autism first made? 8) What type of professionals were involved in giving the eventual diagnosis of autism? 9) What conclusions can be drawn about how children, such as autistic children, who show multiple developmental delays should be referred in order to maximize the efficiency and completeness of the screening and diagnostic process?

METHODS

Subjects

Subjects were 75 children who had been diagnosed as having early infantile autism. Of the 75, 26% had not received a DSM-III diagnosis of autism prior to participation in our research, although these subjects had been described by others as "autistic-like". Therefore, all subjects, regardless of a previous diagnosis of "autism" or "autistic-like", had firm diagnoses made or reconfirmed through the Stanford Autism Project by our multidisciplinary team using a standardized evaluation (Siegel, et al., 1986) and DSM-III criteria before selection for this study. For the child, the standardized evaluation included intelligence testing, observation of the child playing alone, and a standardized play session examining the child's responsiveness to parents and strangers under conditions of high, low and no task demand. The parents gave relevant family and developmental history and records of previous developmental and medical evaluations were reviewed. Subjects ranged in age from 4 years to 25 years (6% older than 20 years) at the time of our evaluation.

Study 1- Chart Review

Approximately 100 charts of autistic children were examined for completeness. A chart was regarded as complete if it contained a full set of evaluations from all agencies named by parents as keeping extensive records on their child (e.g., primary care physician, plus the state developmental services agency, plus the school district). Of these, 49 cases were rated as complete and selected for review. The subjects included 38 males, and 11 females; 8% (N=4) were $\leq$ 5 years old, 47% (N=23) were 6-10 years old, 22.5% (N=11) were 11-15 years old, and 22.5% (N=11) were $\geq$ 16 years old.

Study 2- Parent Survey

To validate and expand the findings of Study 1, parents were surveyed by questionaire about how their children had been diagnosed as autistic. Additional questions were added that were not consistently available through chart review, including when parents first had become concerned, and what they viewed as the strengths and limitations of the diagnostic process. Questionaires were mailed to parents of the 49 subjects of Study 1, plus all new cases of autism that had been added to the research project's series subsequent to Study 1 (N~50). There were 51 respondents; of whom 24 were parents of subjects in Study 1. The 51 subjects included 41 males and 10 females; 14% (N=7) were $\leq$ 5 years old, 37% (N=19) were 6-10 years old, 29% (N=15) were 11-15 years old, and 20% (N=10) were $\geq$ 16 years old. Excluded from Study 2 were those lost to follow-up since the Study 1 charts had been compiled and some parents who decided that it had been too long ago to recall what had happened at the time their child had been diagnosed.

There were 24 cases included in both Studies 1 & 2 (the agreement sample), and these were examined to determine the accuracy of parent recollections by comparing parent survey responses to actual reports in the charts.

RESULTS

The participation of parents and professionals in the diagnosis of our sample can be analyzed in three main stages: 1) what aspects of development parents first became concerned about, 2) the process through which some professional acknowledged this concern--resulting in what is referred to here as the initial diagnosis, and 3) the process through which the child ultimately was diagnosed as having autism--resulting in what is referred to here as the definitive diagnosis.

The results are identified according to whether the data come from Study 1, Study 2, or the agreement sample of the 24 overlapping cases. Group data are expressed as means and standard deviations.

Fig. 1 About Here

First Concerns about Developmental Delays

Parents were surveyed about when they had first become concerned that there might be a problem in their child's development (Study 2). On average, they report (in Study 2) having been concerned by the time the child was 18.3 ± 12.3 months (see Fig. 1). We examined separately reports of first concerns among first-time parents and among parents with older children. Experienced parents reported initial concerns, on average, by 13.7 months; of these, six reported being concerned from the first month of life. First-time parents reported initial concerns later, on average, by 21 months of age.

Parents were asked in which areas of development they had first felt that problems might be occurring (Study 2): 98% cited concerns with language development, 84% cited concerns with social development, and 34% cited concerns with motor milestones. These total more than 100% because parents often reported becoming concerned simultaneously

with more than one area of development. Among these parents, 92% reported that their concerns had been discussed initially with their child's primary care physician (84% with pediatricians, 8% with general or family practitioners). The remaining 8% of parents self-referred directly to other professionals.

Initial Diagnosis

In investigating the age of the child when diagnostic work began, we regarded the initial diagnosis as the first "label" given the child's condition, regardless of accuracy or whether it was a DSM-III or other medical diagnostic category. On average, we found an interval of 13 months between when parents first became concerned about their children's development, and when the children received initial diagnoses. In Study 1, mean age at initial diagnosis was $30^{+}_{-}13$ months old. In Study 2, it was $31^{+}_{-}15$ months old (see Fig. 1). For children in the agreement sample, the mean difference between chart report (Study 1) and parent report (Study 2) for the age at initial diagnosis was only two months. Table 1 shows the variety of initial diagnoses given and the frequency for each. In the agreement sample, parents reported the same initial diagnosis as the charts in 17 out of 19 non-missing cases (89.5% agreement).

Table 1 About Here

We investigated parent's source of initial diagnostic information about their children. First, we asked who had made the initial diagnosis (see Table 2A). Initial diagnoses were most likely to be made by pediatricians or teams. (A team was defined as two or more evaluators from two or more professions who together produced a consensus diagnosis for the child.) Across Studies 1 & 2, about two-thirds of initial diagnoses came from one of

these two sources. In the agreement sample, parents reported the same source for the initial diagnosis as did the charts in 17 out of 19 non-missing cases (89.5% agreement).

One concern of this study was the completeness of the diagnosis that a child with multiple developmental delays such as autism initially received. For example, had the child simply been labeled "delayed", or had the delay had been attributed to autism from the start? In Study 1, 31% received definitive diagnoses of autism, initially. Another 31% received incomplete or nonstandard initial diagnoses, e.g., descriptions such as developmental delay with language and social problems. The remaining 36% received nondefinitive or inaccurate diagnoses, including mental retardation with autism ruled out. Mental retardation was classified as a nondefinitive diagnosis, because a major purpose of diagnosis is treatment planning, and treatment for a child with autism compared to one with mental retardation only, was seen as sufficiently different to warrant this distinction.

In Study 2 (N=48), this pattern was similar, with only 29% of parents reporting that the initial diagnosis was definitive, having been either autism, PDD or "autistic-like". In addition, Study 2 parents were asked whether they first had encountered the term "autism" as a description or as a diagnosis: 41% stated it was first used as a description ("autistic-like", "autistic features"); 59% stated it was first used as a diagnosis. Some parents wrote in comments that they had not encountered the word "autism" before it was applied to their child.

Table 2 About Here

Audiological and neurological examinations were frequently ordered as preliminary steps toward arriving at an initial diagnosis. We examined the yield for positive findings from this approach to diagnosis. Because we were concerned about how well parents

understood and could report on the outcome of such examinations, we first analyzed agreement of Study 2 parent reports compared to records from Study 1. When there was agreement that the test had been made, there was a 96.7% agreement about whether the outcome of an audiological exam had been positive, negative or inconclusive (N=19), and a 97.8% agreement as to whether a neurological exam had yielded positive or negative findings (N=20). Since parent agreement with charts was high, and normal results may have been excluded from our charts more often than remarkable results, we report data on hearing and neurology tests from parent reports given in Study 2 only. In Study 2, 47 out of 48 (97.9%) parents said they remembered that their child had received audiological testing. Of those 47, 43 (91.5%) had hearing sufficient for the development of speech, three (6.4%) had some loss, and testing on one subject (2%) was judged to be inconclusive. Among 47 parents who recalled whether or not their child had been seen by a neurologist, 35 (74.5%) said their child had had such an examination. Of these 35, 26 (74.3%) were judged to have no significant findings, six (17.1%) examinations yielded some positive findings, and three (8.6%) examinations were inconclusive.

Definitive Diagnosis

Typically, a definitive diagnosis came about two years after the initial diagnosis. Specifically, in Study 1, age at definitive diagnosis was $56^{+}_{-}40$ months old. In Study 2, it was $53^{+}_{-}39$ months (see Fig. 1). For children in the agreement sample, there was a mean difference of 6 months in age at definitive diagnosis as calculated from charts versus from parent report; however, there was no difference in median age. This was because a subgroup of parents recalled later ages for definitive diagnoses than actually shown in their children's charts. This finding suggests that some parents may continue to deny their child's diagnosis for some time, even after it has been given to them in a written report.

Fig. 1 shows that 72.5% of children were diagnosed by age five when they entered

school. However, among those not diagnosed by school entry, only a few percent more each year were then diagnosed definitively for the first time. More generally, Fig. 1 shows that most diagnostic evaluation, whether it resulted in an initial or definitive diagnosis, was being carried out on children under five years old.

Finally, we assessed which evaluators had made definitive diagnoses of autism (see Table 2B). Teams were the most likely to make the definitive diagnosis. However, a t-test to determine whether teams diagnosed children earlier than single evaluators showed no significant difference. Although Studies 1 & 2 produced somewhat discrepant findings in this regard, psychologists (Study 1), and psychiatrists (Study 2) were the single evaluators most likely to provide a definitive diagnosis of autism. For children in the agreement sample, there was 91% agreement as to the source of information for the definitive diagnosis (N=23). The most common source of disagreement arose from parents seeing one member of a multidisciplinary team as the sole disseminator of the diagnosis, usually a psychiatrist.

Case Vignette. *Mike.* Mike, a 15-year-old autistic boy, is the youngest of four children born to a 41-year-old mother who had three miscarriages prior to his birth, she had an unidentified "flu" for two weeks in the first trimester of pregnancy, but otherwise pregnancy and delivery were unremarkable. Mike's parents were concerned about him from early infancy because Mike cried almost continually for the first six months of his life. His pediatrician suspected colic, and when Mike calmed down at six months, his parents relaxed somewhat. However, his parents continued to be concerned about his complete lack of responsiveness to them and his siblings and his inconsistent responses to sounds. At 14 months of age, Mike had a febrile seizure, and his pediatrician referred him to a neurologist who diagnosed him as mentally retarded (initial diagnosis). When Mike was 33 months old, his parents sought an evaluation from a multidisciplinary agency which diagnosed him as neurologically handicapped with autistic features. When Mike was four years old, a child psychiatrist saw him as "a very

frightened little boy". When Mike was five, a pediatric neurologist referred him for special education and speech therapy but did not see any other significant problems. Mike entered a private special education program when he turned six, and after two years there, he was first diagnosed as having autism (definitive diagnosis).

DISCUSSION

Age Trends and Identification of Developmentally Multihandicapping Disorders

Many parents had concerns about their children's slow or uneven development in language and socialization by 1-1/2 years of age and expressed these concerns to their pediatricians. When a parent, especially an experienced parent, presents *multiple* developmental concerns, as early as the first year, it may serve as a red flag both for the potential severity of the child's disability, and for the need for screening the extent of possible delays in other areas. Clinically, it is our impression that while pediatricians often are presented with complaints confined to one aspect of development at a time, and these often turn out to be of no concern, complaints made in two or more domains simultaneously are less usual, and may be more worrisome. This study has shown that, in the case of autism, parents typically made reliable observations about simultaneous delays in social and language development when their children were as young as eighteen months old.

One possible contributing factor to the finding that initial diagnoses were not made until around 2-1/2 years of age is the unavailablity of developmentally sensitive screening procedures. Commonly used screening instruments--e.g., the Denver Developmental Screening Test, which assesses perceptual-motor development and more purely cognitive factors, may not be, especially at early ages, as useful as the parents' and pediatrician's own observations for children with uneven development. This is because there may be delays in some domains (e.g., language, social) but not in others (e.g., motor, perceptual). In a developmentally multihandicapped child these tracks of development do not necessarily proceed in tandem. There is a need for developmentally based screening methods that are sensitive to patterns of uneven growth as opposed to global delay. We presently are field-testing such a screening measure for the social and language failures seen in pervasive developmental disorders (Siegel, 1986; Siegel &

Whittaker, in preparation.)

Another possible contributing factor to the lag found between initial parental concern at 1-1/2 years and initial diagnosis at 2-1/2 years is that some parents could not act upon their concern about developmental delay in their child, because of denial. The self-questioning, pain and gradual acceptance that parents experience as they try to figure out how to care for their developmentally disabled child may also account for the two years that typically elapsed between initial diagnosis and definitive diagnosis, which occurred around age 4-1/2 years.

Approximately 75% of subjects had received their definitive diagnosis by age five, when they would have had to enter school. This observation further supports the idea that definitive diagnostic information was seen as relevant to treatment, for it was most often obtained before the child would have been placed in a diagnostically specific school program. By contrast, special education programs before age five are generally more generic than those for children five years and older. Once school was begun at age five, parents may have felt it was less urgent to have their child fully diagnosed, because he was receiving treatment at school. However, we believe that continuing to carry an incomplete or inaccurate diagnosis might be associated with an increased likelihood of a less appropriate educational placement than carrying an accurate diagnosis. As the primary care physician, the pediatrician is probably the best-placed source of professional guidance for a parent whose child might benefit from expeditious completion of the diagnostic process.

One limitation to the present study is that we retrospectively analyzed the diagnostic histories of children spanning a wide age range. Therefore, we can only conjecture that the average of three years that elapsed in this cross-sectional analysis of stages of identification and diagnosis of autism is actually getting shorter because of increasing prevalance of collaborative diagnostic evaluation, a better understanding of the etiology of autism now as compared to ten years ago, and perhaps better parent education about

child development.

Pediatric Management of the Developmentally Multihandicapped Child

Only a few studies address how pediatricians as primary care physicians can best be involved in early identification, diagnosis, and follow-up of children with multiple developmental delays, including those with autism. The roles of the pediatrician can include: 1) administering initial developmental screening, 2) serving as the coordinator of evaluations from other disciplines, and 3) tracking and coordinating interventions.

Screening. Parents usually first approach pediatricians with developmental concerns, so it is valuable for the pediatrician to know when to screen and what to include in screening. Blackman (1986), writing a position paper for the National Center for Clinical Infant Programs, outlined criteria for tracking at-risk infants and toddlers. He pointed to the importance of monitoring different sources of data, including: 1) "persistent, if undefined, professional or parental concern about a child's status", 2) parental and psychosocial factors, 3) health maintenance, chronic illness, and growth failure, 4) familial disorders with developmental implications, 5) central nervous system insult, and 6) atypical or delayed cognitive, socioemotional, motor, sensory or behavioral development.

Freeman & Ritvo (1984) have provided specific developmentally based guidelines for pediatricians screening possibly autistic young children, emphasizing the need for tracking in three areas: 1) sensorimotor development, 2) speech and language, and 3) relating to people, objects and events. They emphasize that organic, cognitive, psychological, family, and social components should be included in evaluations to consider possible etiologies that may include or exclude autism.

Screening by the pediatrician prior to beginning an initial diagnostic evaluation is important in developmentally multihandicapping conditions such as autism as a way of narrowing the field of possibly rare conditions that may need to be ruled out. Schopler

(1978) cited three reasons why autism, in particular, was difficult to diagnose, and these reasons are congruent with the findings of this study: 1) inexperience of the diagnostician, partly because autism occurs infrequently, 2) complexity of the syndrome and the need for a multidisciplinary analysis, and 3) the planned function for the diagnosis (e.g., medication versus school planning). Levy (1983) added a fourth reason--namely that early intervention programs mask symptoms. For example, autistic infants and toddlers who have received a generic infant stimulation program may appear less impaired in such respects as ability to make eye contact than similarly impaired, but untreated patients of the same age. Fifth, the most frequently used paper-and-pencil checklists for autism, lack discriminative validity, failing to exclude other more frequent diagnoses such as severe mental retardation (Parks, 1983). We postulate a sixth reason, that any "developmental" disorder follows a developmental course and is manifested differently according to the child's chronological and mental ages, especially in the first five years of life when identification is taking place.

Coordinating Evaluations. In this study, the pediatrician's role in coordinating evaluations from other disciplines appeared to occur in two ways: 1) the pediatrician referred the child to a multidisciplinary team, or 2) the pediatrician arranged for serial visits to single evaluators, one of whom then arrived at the diagnosis of autism. The findings showed that, when pediatricians followed the first route and children were seen by multidisciplinary teams, the children were more often identified accurately as having autism. The advantages of a multidisciplinary team probably included convergence of different observations from each evaluator and, perhaps, an attitude of "the buck stops here". From a case management viewpoint, a team evaluation was self-tracking, alleviating the need for the pediatrician to be highly involved in interim decision-making about which evaluator should see the patient next.

When pediatricians followed the second route, and patients were serially sent out for consultations with different evaluators, parents often encountered many ruled-out

diagnoses before they were told what was ruled-in. Review of narrative data from the surveys showed that parents experienced a great deal of anxiety during this period between initial and definitive diagnosis when they knew *something* was wrong with their child but did not know what. Of greatest concern to case management are those parents who become so frustrated, confused, or discouraged during this interim period that they fall back on their denial and fail to follow through and complete the diagnostic process.

Tracking interventions. Another role for the pediatrician in managing a developmentally disabled child is tracking and coordinating interventions. Ten years ago, very little intervention was available to a developmentally handicapped infant or toddler. Now such programs are increasingly available and domain specific, e.g. focusing on language, behavior or motor development. In the case of autism, differences in treating it versus mental retardation alone, make the differential diagnosis worth pursuing. For example, although many mentally retarded toddlers respond to social rewards such as praise and affection, autistic toddlers usually need to be offered primary rewards such as food and often do less well in programs where primarily social rewards are used.

An analogy may be drawn between the early detection of childhood leukemia and early identification of autism, both similarly rare disorders. When leukemia was uniformly fatal, detection was less relevant than since the development of effective treatment. Now that interventions for very young, developmentally disabled children are available, proceeding with screening that leads to early accurate diagnosis and appropriate intervention is more critical.

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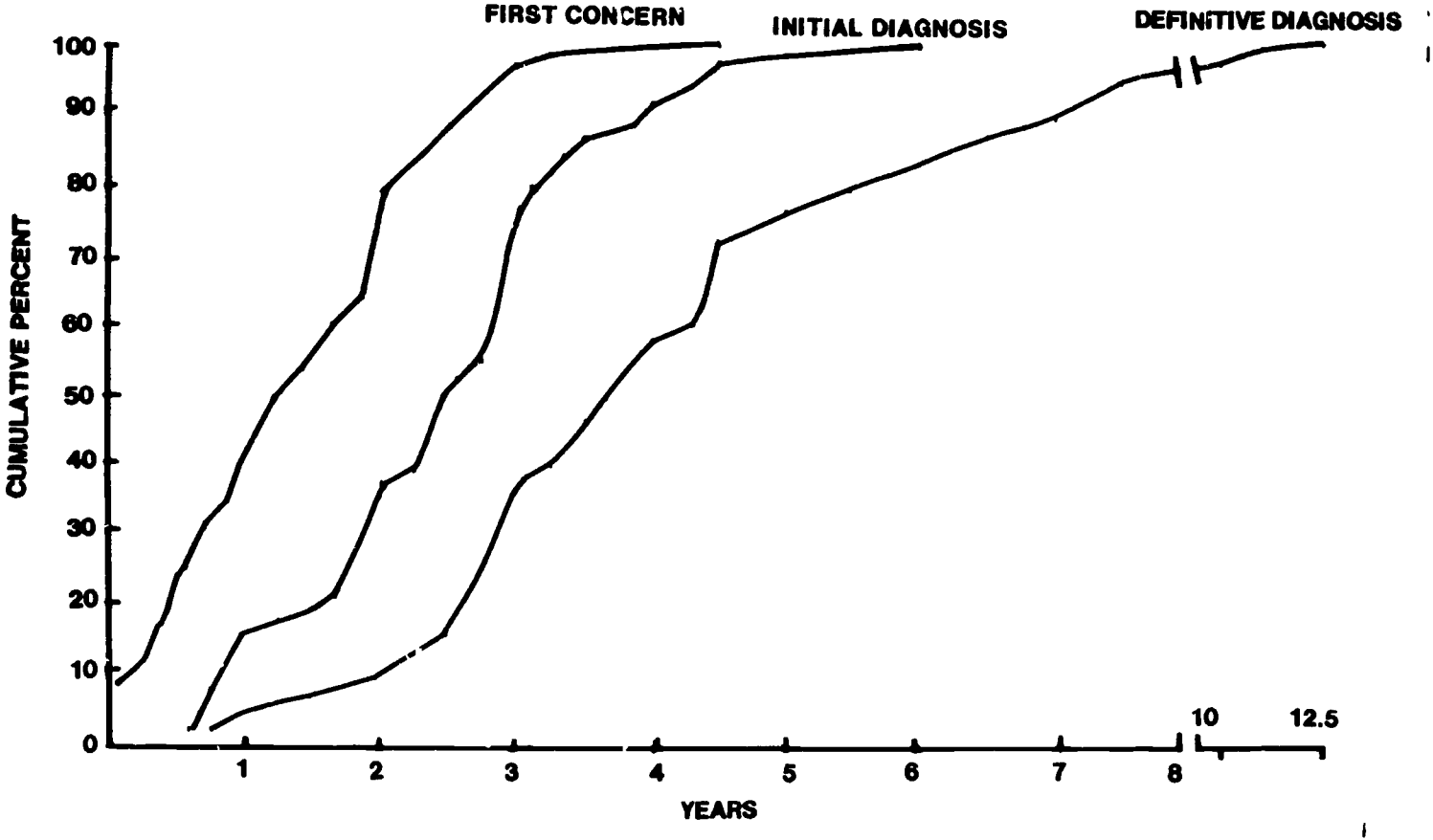

Fig. 1. Cumulative percentages of children for ages at: initial parental concern about delayed development, initial diagnosis, and definitive diagnosis of autism.

TABLE 1.

Initial diagnoses for children with autism

Diagnosis	Study 1 (N=45)	Study 2 (N=51)
Au'ism/Autistic-like/PDD	40%	34%
Mental Retardation Developmental Delay	31%	33%
Neurological Disorder	7%	6%
Normal	7%	6%
Language Delay	4%	2%
Other: Anti-social, Psychotic, not Autism	11%	18%

TABLE 2.

Sources of diagnostic information for autism/PDD

	A. Initial Diagnosis		B. Definitive Diagnosis	
Diagnosed by:	Study 1 (N=45)	Study 2 (N=51)	Study 1 (N=49)	Study 2 (N=51)
Multidisciplinary Team	30%	30%	59%	53%
Pediatrician	29%	37%	6%	4%
Neurologist	14%	13%	10%	4%
Psychologist	8%	2%	16%	4%
Psychiatrist	4%	4%	4%	24%
Audiologist/ Speech Pathologist	4%	4%	2%	0%
Special Educator	0%	0%	2%	6%
Other: Nurse, Friend, Relative, etc.)	11%	10%	1%	4%